

Birth Prevalence of Skeletal Dysplasias

The prevalence of skeletal dysplasias at birth has received relatively little attention, and the completeness of the available data has been viewed with concern. Two recently reported prospective, population-based studies shed light on this subject.

Stoll and colleagues examined birth records, roentgenographic reports, autopsy reports, follow-up pediatrician notes, and other available data from 11 maternity hospitals where all births were recorded from Strasbourg, France and the surrounding region, from 1979 to 1986. The data from fetuses delivered with a minimum age of 20 weeks and from pregnancies interrupted following prenatal diagnosis of a skeletal dysplasia were included; ascertainment was thought

to be complete. A skeletal dysplasia was diagnosed in 34 cases out of 105,374 births to give a prevalence rate of 32.2 per 100,000. The rates per 100,000 births for several of the more common disorders were: achondroplasia, 6.4; thanatophoric dysplasia, 2.8; achondrogenesis, 2.8; osteogenesis imperfecta, 6.4; osteopetrosis, 1.8; and multiple exostoses, 1.8. Roughly half of the patients had disorders that are usually lethal in the newborn period.

The second study, by Andersen, examined the birth prevalence of lethal bone dysplasias. Clinical and radiographic findings were analyzed from all births, including stillbirths, in the county of Fyn, Denmark, from 1970 to 1983. Twelve

lethal bone dysplasias were diagnosed out of 77,977 total births to give a prevalence of 15.4 per 100,000. Three cases of thanatophoric dysplasia (including one with cloverleaf skull) and five cases of achondrogenesis type II were identified, yielding respective prevalence values of 3.8 and 6.4 per 100,000 births, respectively.

Stoll C, Dott B, Roth M-P, et al. *Clin Genet* 1989;35:88-92.

Anderson PE. *Am J Med Genet* 1989;32:484-489.

Editor's comment—*These reports provide relatively similar birth prevalence rates for skeletal dysplasias when one takes into account that the study by Stoll and colleagues examined both lethal and nonlethal conditions whereas*

Andersen looked only at lethal disorders. Together the data suggest that the prevalence of all skeletal dysplasias is slightly greater than 30 per 100,000 births, of which approximately half are lethal in the perinatal period. This figure translates into a rate of around 1 per 3,000 births. This rate, which is actually an underestimate as many disorders are not evident at birth, is much higher than is generally ap-

preciated. For comparison, approximate prevalence rates for several well-known genetic conditions are as follows: Down syndrome, 1 per 700 births; cystic fibrosis, 1 per 1,600 births; muscular dystrophy, 1 per 3,500 male births; hemophilia, 1 per 5,000 male births; phenylketonuria, 1 per 15,000 births; and albinism, 1 per 40,000 births.

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